

RATHKE'S CLEFT CYST AND PARTIAL FEET ADACTYLY

An unusual association

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ABSTRACT - A 53 year-old woman presented a recurrent bifrontal headache of 2 years duration and bilateral progressive visual disturbance. The clinical and neurological examination showed a bilateral feet adactyly and bitemporal hemianopsia. The brain MRI demonstrated a Rathke's cleft cyst. The patient was operated by a transnasal endoscopic approach. It seems that this unusual association has never been described before.

KEY WORDS: adactyly, endoscopic, pituitary surgery, pituitary tumor, rathke's cleft cyst, transnasal approach.

Cisto de Rathke e adactilia parcial dos pés: uma associação rara

RESUMO - Mulher de 53 anos com história recorrente de cefaléia com duração de 2 anos bilateral e progressiva, acompanhada de distúrbios visuais. O exame clínico e neurológico mostrou uma adactilia dos pés e hemianopsia bitemporal. A ressonância nuclear magnética cerebral mostrou um cisto de Rathke. A paciente foi operada por via transnasal endoscópica. Aparentemente esta é a primeira vez que esta associação é descrita na literatura.

PALAVRAS-CHAVE: adactilia, endoscopia, cirurgia hipofisária, cisto de rathke, tumor de hipófise, via transnasal.

Rathke's cleft cyst (RCC) is an epithelial cell-lined cyst of the pituitary believed to derive from remnants of Rathke's pouch, a dorsal invagination of the stomodeal ectoderm¹. Partial adactyly is an autosomal congenital anomaly of the hand or foot, marked by persistence of the webbing between adjacent digits.

We report an unusual association of these two entities in a 53 years old woman.

CASE

A 53-years-old woman with an unremarkable family history, issued of not consanguineous parents, presented a recurrent bifrontal headache of 2 years duration and bilateral progressive visual disturbance. Endocrinological evaluation included measurement of baseline levels of ACTH, cortisol, TSH, free T₃, free T₄, GH, PRL, LH, FSH, and oestradiol. Dynamic stimulation test were done with insulin (0.1 U/kg), TRH (500 µg), GnRH (100 µg) CRH (100 µg) and GHRH (100 µg). The patient had no primary disturbance of thyroid, adrenal and gonadal functions. Her physical examination showed bilateral partial adactyly of the feet (Fig 1) and the ophthalmologic examinations showed bitemporal hemianopsia. Chromosomes were apparently normal. Neuroimaging (MRI) revealed a Rathke's cleft cyst (20x15x14 mm) with intra and supra sellar components (Figs 2 and 3). The patient

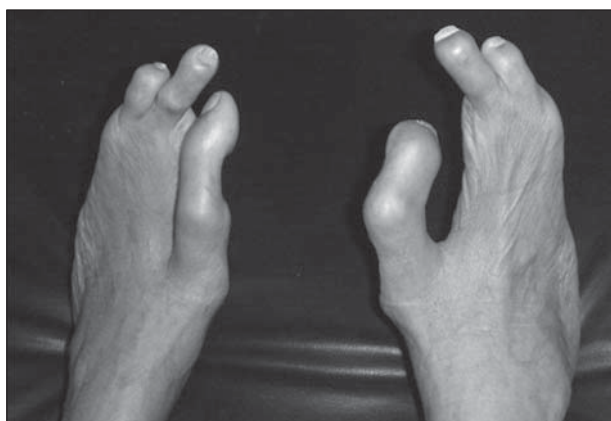


Fig 1. Bilateral foot fusion. Partial adactyly.

underwent a transnasal endoscopic surgery with drainage and resection of the cyst wall (Figs 4 and 5). The last examination of the patient two years after neurological surgery showed a complete recuperation of the hemianopsia, and the orthopedic surgery service has done a partial corrections of her feet adactyly.

DISCUSSION

Rathke's cleft cysts are a benign epithelium-lined intrasellar cysts believed to originate from remnants

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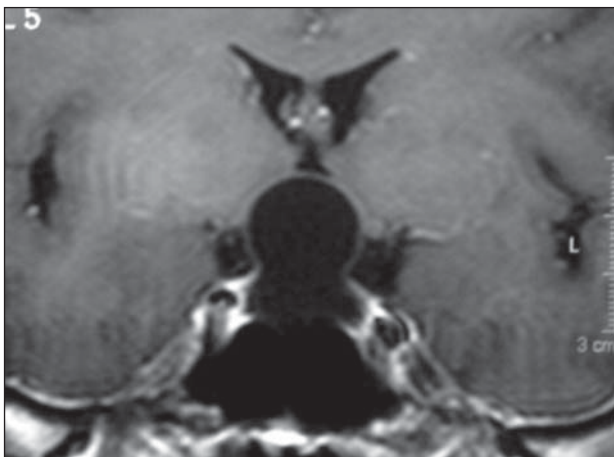


Fig 2. Postgadolinium coronal T1-weighted MRI scan showing a low intensity Rathke's cleft cyst.

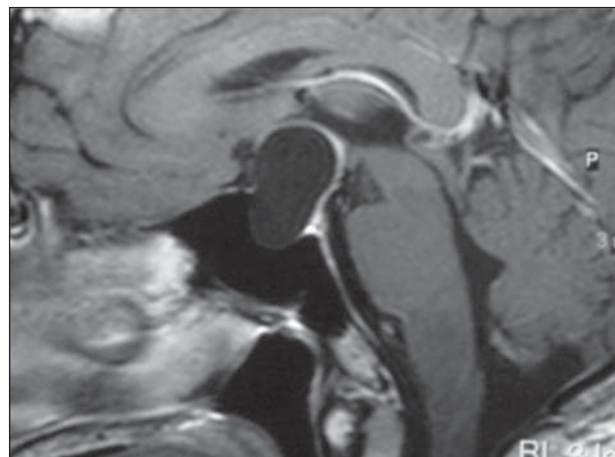


Fig 3. Postgadolinium sagittal T1-weighted MRI scan showing a low intensity Rathke's cleft cyst.

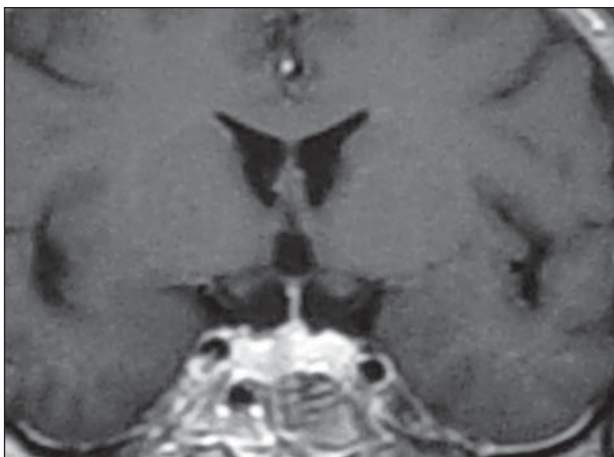


Fig 4. Post operative postgadolinium coronal T1-weighted MRI scan showing no more Rathke's cleft cyst.

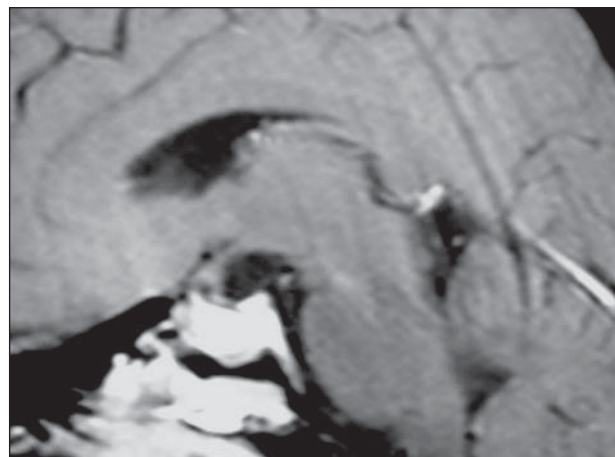


Fig 5. Post operative postgadolinium sagittal T1-weighted MRI scan showing no more Rathke's cleft cyst.

of the Rathke pouch, a dorsal invagination of the stomodeal ectoderm¹. RCCs commonly have a round, ovoid or dumbbell shape and rarely cause symptoms. They have been reported in 2-26% of pituitary glands studied at autopsy² or in as many as 33% of cases³. No racial predilections recognized and there is a male-to-female ratio of 1:2. The patient's age at presentation ranges from 4-73 years (mean age, 38 years). The highest frequency is in those aged 50-60 years³. On MRI RCC usually does not exhibit destruction or enlargement of the sella turcica⁴, but there is in our case. Intracystic nodules, that are waxy nodules, if present, show characteristically intensity on MRI⁵. RCCs are almost always homogeneous in MR intensity, except for waxy nodules, whereas other lesions such as cystic craniopharyngioma and hemorrhagic adenomas are less frequently homogeneous⁶. A high T1-WI intensity has been interpreted to indicate a

high content of protein and mucopolysaccharide and, rarely are hemorrhage⁷. The cystic content of high and isointensity RCCs on T1-WI is usually mucus with varying viscosity. By contrast, RCCs with low intensity on T1-WI usually contain CSF-like, transparent fluid, with low viscosity⁸.

Syndactyly (webbed toes or fingers) occurs in approximately one in 2,000 to 2,500 live births. There are various levels of syndactylization, from partial to complete. The most frequent site is between the second and thirds toes. Syndactyly is thought to be genetic, with an autosomal dominant pattern of inheritance. Cleft foot (lobster foot) is an anomaly in which a single cleft extends proximally into foot, sometimes even as far as midfoot. Generally one or more toes and parts of their metatarsals are absent, and often the tarsals are abnormal. Although the deformity varies in degree and type, the first and fifth

toes usually are present. If a metatarsal is partially or completely absent, its respective toe is always absent too⁹. Recently some authors have connected cigarette smoking during pregnancy and an elevated risk of having a child with polydactyly, syndactyly, or adactyly. Differential diagnosis can be considered with: Apert syndrome (a rare autosomal dominant disorder characterized by craniosynostosis, craniofacial anomalies, and severe symmetrical syndactyly cutaneous and bony fusion of the hands and feet), Saethre-Chotzen syndrome (a relatively mild form of acrocephalosyndactyly with a variable pattern of craniofacial digital, and bone abnormalities), Pallister-Hall syndrome (central and postaxial polydactyly, hypothalamic hamartoma, bifid epiglottis, imperforate anus, renal abnormalities, pulmonary segmentation and inherited in an autosomal dominant pattern), Ellis-van Creveld syndrome (congenital heart defects, polydactyly, multiple frenula, and natal teeth), Smith Lemli-Opitz syndrome (polydactyly and various SNC anomalies), oral-facial-digital syndrome type VI (autosomal recessive polydactyly, tongue hamartoma, and cerebellar vermis hypoplasia), Kaufman-McKusick syndrome (autosomal recessive, hydrometrocolpos, polydactyly, and congenital heart defects), and Greig cephalopolysyndactyly syndrome (autosomal dominant, polydactyly, and craniofacial abnormalities)¹⁰⁻¹³.

In conclusion, histological verification confirmed a Rathke's cleft cyst. Two years after surgery the patient is well and recuperated her vision. The association of Rathke's cleft cyst and feet adactyly in a 53-year-old woman is an unusual association. In an exhaustive literature review it seems to be the first reported case.

We propose that this patient may represent a clinically and perhaps genetically distinct entity, based on normal survival, normal intelligence, lack of endocrine dysfunction or facial anomalies, and no other structural malformation.

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